Congenital Hypothyroidism

Congenital hypothyroidism (CH) occurs in babies who do not have the ability to produce adequate amounts of thyroid hormone. Most cases are sporadic, but it occasionally occurs in siblings and may be inherited as an autosomal recessive disorder. The most common causes of primary hypothyroidism include: thyroid gland aplasia or hypoplasia; ectopic thyroid gland; or enzyme deficiencies in thyroxine (T4) synthesis. Less commonly, hypothyroidism is induced by medications (antithyroid drugs or excess iodine) in the mother. Newborn screening also detects secondary hypothyroidism resulting from failure of the pituitary gland to release thyrotropin (TSH), and tertiary hypothyroidism resulting from failure of the hypothalamus to secrete thyrotropin-releasing hormone (TRH).

Estimated Incidence (MI):	1:2,000
Laboratory Screening Test:	Fluorometric assay screens for TSH (thyrotropin)
Timing of Test:	≥24 hours of age: Results are valid False positive results can occur on specimens obtained before 24 hours of age due to the normal physiologic TSH surge that occurs after birth.
Feeding Effect:	None
Transfusion Effect:	None
Confirmation:	All strong and borderline positive tests are referred to the Endocrine Follow-up Program (EFUP) at the University of Michigan (734) 647-8938. The EFUP coordinates follow-up for infants with suspected CH. Diagnosis and treatment is provided through a network of pediatric endocrinologists throughout the state.
Treatment:	Oral thyroid hormone (replacement) administered daily. This should begin as soon as possible after confirmation of the diagnosis.
Comment:	**Most, but not all cases of severe, early on-set hypothyroidism are detected by newborn screening. The screening program is not designed to detect late-onset, clinically moderate or sub-clinical forms of hypothyroidism. The newborn screen should no be relied on to rule out all abnormalities of thyroid function.**

For further information, contact the Newborn Screening Program
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